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Bax基因P53结合位点多态性与食管鳞癌易感性的关联研究

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Title: Association between SNP in P53-binding site of Bax and risk of esophageal squamous cell carcinoma in southwest Chinese Han population

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关键词: Bax; P53; 单核苷酸多态性 (SNP); 食管鳞状细胞癌; 危险因素

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摘要: 目的 探讨Bax基因P53结合位点多态性与我国西南地区食管鳞状细胞癌(esophageal squamous cell carcinoma, ESCC)发病的关联性。 方法 利用SNaPshot技术检测201例食管鳞癌病例组及183例健康对照的Bax基因rs1009316位点多态性的分布情况,分析不同基因型与食管鳞癌易感性的关联。 结果 食管鳞癌病例组和对照组中rs1009316的CC和CT/TT基因型频率分别为59.2%、40.8%和73.2%、26.8%,CT/TT相比CC增加食管鳞癌的发病风险(OR值为1.884,95%CI为1.224~2.901),T等位基因增加食管鳞癌的发病风险(OR值1.620,95%CI为1.103~2.379)。分层分析发现,无吸烟史群体中,T等位基因(OR值为1.858,95%CI为1.050~3.288)及CT/TT基因型(OR值为2.178,95%CI为1.143~4.147)分别增加食管鳞癌的发病风险。 结论 在中国西南地区人群中,Bax基因P53结合区域的多态性位点rs1009316是食管鳞癌的危险因素之一。

Abstract: Objective To evaluate the association between single nucleotide polymorphism (SNP) in P53-binding site of Bax and the risk of esophageal squamous cell carcinoma (ESCC) in southwest Chinese Han population. Methods The SNaPshot technique was applied to detect genotype of rs1009316 in 201 cases of ESCC (ESCC group) and 183 cases of healthy controls (control group), and the association between different genotypes of rs1009316 and the risk of ESCC was analyzed. Results The frequencies of the rs1009316 variants CC and CT/TT were 59.2% and 40.8% in the ESCC group and 73.2% and 26.8% in the control group, respectively. Compared with CC genotype, CT/TT genotype increased the risk of ESCC (OR 1.884, 95% CI 1.224 to 2.901). T allele frequency was 20% in the ESCC group and 14% in the

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control group, and T allele was positively correlated with the risk of ESCC (*OR* 1.620, 95% *CI* 1.103 to 2.379). In stratification analysis, CT/TT genotype and T allele increased the risk of ESCC in the subjects without smoking history.

Conclusion rs1009316 a SNP lying in the p53-binding site of Bax, is a risk factor of ECSS in southwest Chinese Han population.

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